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Brief Clinical Report

Intestinal Malrotation in a Child With Cardio-Facio-Cutaneous Syndrome

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We present a child with cardio-facio-cutaneous (CFC) syndrome with inadequate weight gain due to inadequate food intake. After correction of hyperemesis due to intestinal malrotation, she continued to fail to feed due to poor suck reflex. A review documented digestive system findings in 26 of 57 reported patients with CFC syndrome. Thus, digestive system dysfunction and malformation may represent an additional manifestation of the CFC syndrome. Am. J. Med. Genet. 70:284–286, 1997.

KEY WORDS: abnormal face; cardiac defects; abnormal hair and skin; mental retardation syndrome; growth failure; multiple congenital anomalies; malrotation; digestive system dysfunction

INTRODUCTION

The cardio-facio-cutaneous (CFC) syndrome [Reynolds et al., 1986] comprises congenital heart defects, a characteristic facial appearance, abnormal hair and skin, and psychomotor and growth retardation. Krajewska-Walasek et al. [1996] reported on 2 new cases with a review of 46 known cases to give a detailed clinical description of the syndrome, citing 8 cases of hernia and 11 cases of hepato- or splenomegaly as additional common findings. This is a report of a child with CFC syndrome, intestinal malrotation, and severe feeding problems.

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CLINICAL REPORT

XJ was born at 38 weeks of gestation to a 28-year-old gravida 2 para 1 mother and a 27-year-old father, both of Hispanic origin. Their union is nonconsanguineous. The pregnancy was complicated by polyhydramnios at 7 months of gestation. The infant weighed 2,757 g (25th centile), was 50 cm long (75th centile), and had an OFC of 34.5 cm (75th–90th centile). She was hospitalized for 1-1/2 months for inadequate weight gain and frequent postprandial vomiting. Intestinal malrotation was suspected but not confirmed by 2 upper gastrointestinal (GI) series. She was treated with ranitidine hydrochloride and Propulsid® syrups and her formula was changed to Nutramigen®, providing some improvement. Cardiac evaluation showed patent ductus arteriosus (PDA) and pulmonary stenosis. Other neonatal problems were transient hypocalcemia and extensive monilial skin rash on the neck and shoulder. The skin became hyper- and hypopigmented after the monilial infection was cleared by nystatin.

She was seen at age 2-1/2 months for evaluation of her unusual facial appearance. She weighed 3.4 kg (<5th centile), was 53 cm long (<5th centile), and had an OFC of 37.25 cm (10th centile). She had relative macrocephaly, sparse curly-woolly hair, high forehead, bitemporal constrictions, hypoplasia of the supraorbital ridge with thin eyebrows, sparse eye lashes, down-slanting palpebral fissures, strabismus, depressed nasal bridge, anteverted nostrils, long philtrum, apparently low-set ears with prominent helices, and a highly arched palate (Fig. 1). A grade III/VI systolic cardiac murmur was heard. There were hyper- and hypopigmented skin lesions on the neck from chin to shoulder.

Chromosome analysis showed a normal female karyotype of 46,XX. In situ hybridization studies using a D22S75 DiGeorge chromosome probe and a D22S39 chromosome 22 probe (Oncor) were done because of hypocalcemia, congenital heart disease, and feeding problems. The results showed 2 signals in both chromosome 22; no del (22) (q11.2) was found. Cardiac evaluation at this time showed normally closed PDA and moderate pulmonary stenosis.

GI evaluation showed malrotation of the small intes-

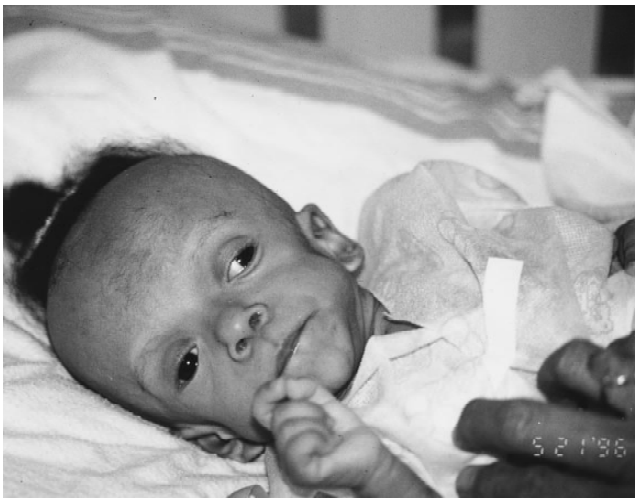

Fig. 1. The patient at age 6 months.

tine which was corrected. After the operation, attempts to feed orally were unsuccessful due to the child's inability to suck adequately, and a gastrostomy tube was placed. At 8 months, she weighed 4.16 kg, had a social smile, but was not able to roll over.

Both parents and the older brother have straight hair. Family history is unremarkable.

DISCUSSION

Our patient has the CFC syndrome on the basis of characteristic facial anomalies, sparse curly-woolly hair, skin involvement, pulmonary stenosis, psychomotor and growth retardation, and apparently normal chromosomes. The severe growth retardation may suggest a diagnosis of Costello syndrome, which has many overlapping features with the CFC syndrome. However, our patient demonstrates the facial traits of CFC syndrome, the salient characteristic that distinguishes between these 2 diagnoses [Zampino et al., 1993]. Additionally, XJ has pulmonary stenosis, which is frequently seen in the CFC syndrome but rarely in the Costello syndrome.

Our patient also had hyperemesis secondary to intestinal malrotation. A review did not uncover other cases that presented with intestinal malrotation, but there were numerous reports which included digestive system findings. There were 57 previously published cases [Ades et al., 1992; Baraitser and Patton, 1986; Borradori and Blanchet-Bardon, 1993; Bottani et al., 1991; Cantú et al., 1982; Chrzanowska et al., 1989; Corsello and Giuffrè, 1991; Dunya et al., 1993; Fryer et al., 1991; Fryns, 1992; Ghezzi et al., 1992; Gross-Tsur et al., 1990; Kajii et al., 1992; Krajewska-Walasek et al., 1996; Leichtman, 1996; Lopez-Rangel et al., 1993; Matsuda et al., 1991; Mucklow, 1989; Neri et al., 1987; Piérard et al., 1990; Raymond and Holmes, 1993; Reynolds et al., 1986; Somer et al., 1992; Sorge et al., 1989; Turnpenny et al., 1992; Verloes et al., 1988; Ward et al., 1994; Young et al., 1993]. As shown in Table I, digestive system manifestations were reported in 27 patients, including the present case. The most frequent finding was feeding difficulty usually associated with

TABLE I. Summary of Digestive System Manifestations in 27 of 58 Reported Cases of CFC Syndrome

Reference	Case	Sex (M:F)	Feeding problems	Spleno megaly	Hepato megaly	Umbilical hernia	Inguinal hernia	Other
Ades et al. [1992]	SA	M		+[a]	+	+		
Baraitser and Patton [1986]	1	F	+					
Cantú et al. [1982]	A	F			+			
Chrzanowska et al. [1989]	FA	F				+		
Corsello and Giuffrè [1991]	1	M					+	
Fryer et al. [1991]	AP	M	+					
Fryns [1992]	–	F	+			+		
Kajii et al. [1992]	YY	M	+					
Leichtman [1996]	1	F	+					
Matsuda et al. [1991]	2	M	+					
Mucklow [1989]	VR	F		+	+			
Neri et al. [1987]	AS	M	+			+	+	
Neri et al. [1987]	PS	M		+				
Reynolds et al. [1986]	1	F				+		
Reynolds et al. [1986]	2	F		+				
Reynolds et al. [1986]	3	M		+				
Reynolds et al. [1986]	4	M					+	
Reynolds et al. [1986]	5	F		+		+		
Reynolds et al. [1986]	6	F	+					
Reynolds et al. [1986]	8	M		+				
Somer et al. [1992]	ML	F	+					
Somer et al. [1992]	TA	F	+			+		
Somer et al. [1992]	SP	F	+	+	+			Anal stenosis
Turnpenny et al. [1992]	–	F	+					
Ward et al. [1994]	2	F	+					
Young et al. [1993]	1	F	+					
Present case	XJ	F	+					Malrotation
Total	27	10:17	15	8	4	7	3	2

[a]Subsequently resolved.

hyperemesis, reported in 15 patients. In 3 other patients, failure to thrive in infancy was reported without reference to feeding difficulty. Additional digestive system findings were apparent splenomegaly (8 cases), hepatomegaly (4), umbilical hernia (7), inguinal hernia (3), anal stenosis (one), and intestinal malrotation (one). Five of the 15 patients with feeding problems had one or 2 of these additional digestive system findings.

In previous reviews, digestive system dysfunction or malformation was not consistently addressed as a manifestation of CFC syndrome. For this reason, some authors may have excluded these findings from their case report, especially when their focus was on one specific manifestation of the syndrome. If so, the true incidence of digestive system manifestation in patients with CFC syndrome may be even higher than what has been reported.

Digestive system dysfunction or malformation in patients with CFC syndrome, especially in infancy, can result in inadequate nutrition and growth retardation. Recognition of this potential problem will facilitate management with assisted feeding procedures such as gastrostomy tube placement, and may reduce the severity of growth retardation.

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